Synopsis in MEDICAL GENETICS for the academic year 2019-2020

1. Methods of human genetic study. Pedigree construction: symbols, pedigree drawing, assessment. Twin study.
2. Methods of human genetic study. Molecular-genetic methods. Principles and approaches for DNA analysis.
   Direct DNA analysis – principles, techniques and application.
3. Methods of human genetic study. Molecular-genetic methods. Principles and approaches for DNA analysis. Indirect DNA analysis – principles, techniques and application.
4. Methods of genetic study. Cytogenetic methods – materials, steps in cytogenetic preparation (banding staining techniques), clinical indication for analysis.
5. Chromosomal mutations – types (numerical and structural), mosaicism, mechanism, clinical significance, recurrence
   risk.
6. Chromosomal disorders – autosomal chromosomal abnormalities (numerical and structural): incidence; general
   clinical characteristics, mechanism, recurrence risk, cytogenetic variants, examples of chromosomal syndromes.
7. Chromosomal disorders – sex chromosomal abnormalities: incidence; general clinical characteristics, mechanism, recurrence risk, cytogenetic variants, examples of chromosomal syndromes.
8. Chromosomal disorders – incidence (spontaneous abortions, stillbirths, newborns), general clinical characteristics (autosomal and sex chromosomal); role of chromosomal abnormalities for human infertility.
9. Pattern of inheritance – autosomal dominant: characteristics of pedigree, role of the specific factors, genetic risks,
   clinical features, examples - Huntington disease, Myotonic dystrophy, Neurofibromatosis and etc.
10. Pattern of inheritance – autosomal recessive: characteristics of pedigree, role of the specific factors, genetic risks,
    clinical features, examples - Cystic fibrosis, β-Thalassaemia, Spinal muscular atrophy and etc.
11. Pattern of inheritance – X-linked (dominant and recessive): characteristics of pedigree, role of the specific factors, genetic risks, clinical features, examples - Duchenne muscular dystrophy, Haemophilia – A and B,
    Fragile X syndrome, Familial hypophosphatemic /vitamin D-resistant rickets.
12. The inborn errors of metabolism (IEM) – prevalence, inheritance, biochemical basis of IEM, common clinical features, examples.
13. The inborn errors of metabolism. Newborn screening program - Mass and selective newborn screening (criteria, requirements, study methods, indications)
14. Multifactorial inheritance. The liability / threshold model. Recurrence risk in multifactorial disorders. Increasing risk factors. Genetic counseling.
15. Multifactorial inheritance. Genetic susceptibility to common disorders. Heritability and recurrence risk. Common multifactorial disorders in adult, examples.
16. Multifactorial inheritance. Genetic susceptibility to common disorders. Heritability and recurrence risk. Common multifactorial congenital anomalies in newborn babies.
17. Dysmorphology. Genetics and congenital anomalies – malformation, deformation and disruption. Definition and examples. Etiology, incidence and clinical significance.
18. Dysmorphology. Genetics and congenital anomalies – syndrome, association, sequence. Definition and examples. Etiology, incidence and clinical significance.

19. Dysmorphology and teratogenesis. Major steps of syndrome identification process. Environmental teratogens - examples.
20. Classification of genetic diseases. Main groups of genetic disorders: characteristics, incidence and significance for human pathology.
21. Organization of human genome. Chromosomes and genes - structure and function. Mutations as a molecular base of genetic disorders.
22. Haemoglobinopathies. Disorders of haemoglobin structure – types of mutations, general clinical features. Examples - Sickle cell disease.
23. Haemoglobinopathies. Disorders of haemoglobin synthesis – types of mutations, general clinical features. Examples α- and β -Thalassaemia.
24. Inherited immunodeficiency disorders. Primary (defects at specific stages of differentiation of stem cells) immunodeficiency – examples.
25. Inherited immunodeficiency disorders. Secondary (associated) immunodeficiency disorders– examples.
26. Unusual pattern of inheritance. Genomic imprinting and uniparental disomy. Examples: Prader–Willi syndrome, Angelman syndrome
27. Unusual pattern of inheritance. Anticipation and triplet repeat expansion. Examples: Huntington's disease, Myotonic dystrophy, Fragile X syndrome.
28. Unusual pattern of inheritance. Mosaicism. Cytoplasmic (mitochondrial) inheritance. Examples.
29. Genetic heterogeneity. Allelic and locus heterogeneity. Examples.
30. Genetic heterogeneity. Pleiotropy. Variable expression and reduced penetrance. Examples.
31. Prevention of genetic disorders – main approaches and levels of organization /primary, secondary/.
32. Approaches for prevention of genetic disorders. Genetic screening (types). Maternal serum screening.
33. Approaches for prevention of genetic disorders. Genetic counseling – definition, aims, main steps, indications.
34. Genetic counseling in different type of genetic disorders – chromosomal, single gene, polygenic/multifactorial.
35. Prenatal diagnosis – main techniques /invasive and noninvasive/, diagnostic methods, optimal time in gestation, associated risk. Indications for prenatal diagnosis.
36. Prenatal diagnosis. Non invasive techniques - optimal time in gestation, diagnostic methods, associated risk, indications.
37. Prenatal diagnosis. Invasive techniques - optimal time in gestation, diagnostic methods, associated risk, indications.
38. Cancer genetics. Tumor suppressor genes (Retinoblastoma, Wilms' tumor).
39. Cancer genetics. Oncogenes (Examples).

Cancer genetics. Mendelian disorders with strong predisposition to cancer. Common cancers: breast and colon cancers.